

Achalasia and hyperthyroidism: A co-occurrence revealing autoimmune connections

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ABSTRACT

Achalasia is a rare esophageal motility disorder characterized by an unknown etiology, myenteric inflammation, and autoimmune markers. We present a case of a 43-year-old female with progressive dysphagia, regurgitation, generalized weakness, and hair loss. Despite normal cranial nerves and normal laboratory values, upper GI endoscopy revealed pangastritis, while manometry indicated type 2 achalasia cardia. The patient underwent laparoscopic Heller's myotomy for achalasia and received treatment for hyperthyroidism. The discussion explores the autoimmune aspects of achalasia and its rare association with hyperthyroidism. Early diagnosis and management are crucial for improving patient outcomes.

Keywords: achalasia, esophageal motility disorder, autoimmune disease, hyperthyroidism

INTRODUCTION

Achalasia is an esophageal motility condition with unknown etiology [1]. Myenteric inflammation is a common and early pathologic alteration in achalasia patients [2]. Though achalasia can appear at any age, the disorder is commonly seen in both sexes in their fifth and sixth decades of life [3]. The finding that achalasia is linked to the HLA-DQw1 gene and that affected patients frequently have circulating antibodies against enteric neurons raises the possibility that achalasia is an autoimmune disease [4]. Significant infiltration of the myenteric plexus by monocytes and antibodies to myenteric neurons further consolidate the case [5]. Many cases of autoimmune thyroid disorders (AITDs) and other autoimmune diseases have been described. A quarter of all individuals with achalasia have a concomitant thyroid condition, most commonly hypothyroidism. Despite its rarity, the link between achalasia and hyperthyroidism deserves consideration [5].

CASE REPORT

A 43-year-old female presented with difficulty in swallowing both solids and liquids for 3 months, insidious and progressive, along with episodes of re-

gurgitation of meals immediately after consumption. She refused any history of fever, painful swallowing of food, or bowel, or bladder complaints. She had associated generalized fatigue, a history of weight loss, a history of hair loss, and occasional unprovoked palpitations. She refused any pattern of fatigable weakness, difficulty in chewing food, or drooping of eyelids. She denied any breathlessness on exertion, chest pain, orthopnea, or Paroxysmal nocturnal Dyspnea. Her complaints had no diurnal variations.

On examination, her BP was 130/70 mmHg, PR: 90/min regular, RR: 18/min, afebrile. On systemic examination, cranial nerves were normal, Gag reflex was present on both sides, and power was 5/5 on all four limbs. Her ECG showed no significant ST-T changes, and chest X-ray was not contributory. An upper GI endoscopy was done to rule out any mechanical causes of dysphagia, which showed no features to suggest any mechanical causes for dysphagia and revealed pangastritis. The MRI brain was done to rule out any central causes for dysphagia and had no significant abnormalities. After ruling out mechanical and CNS causes, we went ahead with the acetylcholine receptor antibody testing to rule out neuromuscular junction disorders like Myasthenia, which came out to be negative. Barium swallow was at-

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tempted thrice to rule out motility disorders and was unsuccessful. She was further evaluated for any thyroid disorders and showed a TSH of <0.015 , FT3: 6.01 FT4: 3.14, and Anti-TPO antibodies positive.

After initially attributing her symptoms to thyroid dysfunction, she was started on Tab Carbimazole 10 mg BD, and she was fed with a Ryle's tube feed given her dysphagia. Her symptoms improved but persisted even on treatment. With a high suspicion of Motility disorder, and previous unsuccessful attempts to Barium swallow, repeated attempts to barium swallow were made and it showed features suggestive of achalasia cardia (Figure 1) which prompted a manometry study. Manometry showed a high basal LES pressure, and high median integrated relaxation pressure (IRP), along with panesophageal pressurization, suggestive of type 2 achalasia cardia (Figure 2).

The appearance of the esophagus in achalasia is known as the "bird-beak sign". If the shape of the narrowed region is irregular and/or involves more than simply the distal esophagus, the alternative diagnosis of esophageal cancer (pseudo achalasia) should be investigated.

The baseline pressure at the upper (UES) and lower (LES) esophageal sphincters is represented by continuous red/orange/yellow lines near the top and bottom of the image. In response to a swallow, the remaining esophageal muscles contract, resulting in a dark region that indicates increased pressure. Instead of the diagonal form that regular peristalsis



FIGURE 1. Barium swallow study - The smooth funnel-shaped tapering (green arrow) of the lower esophageal sphincter is accompanied by the broadening (arrow) of the prestenotic distal esophagus

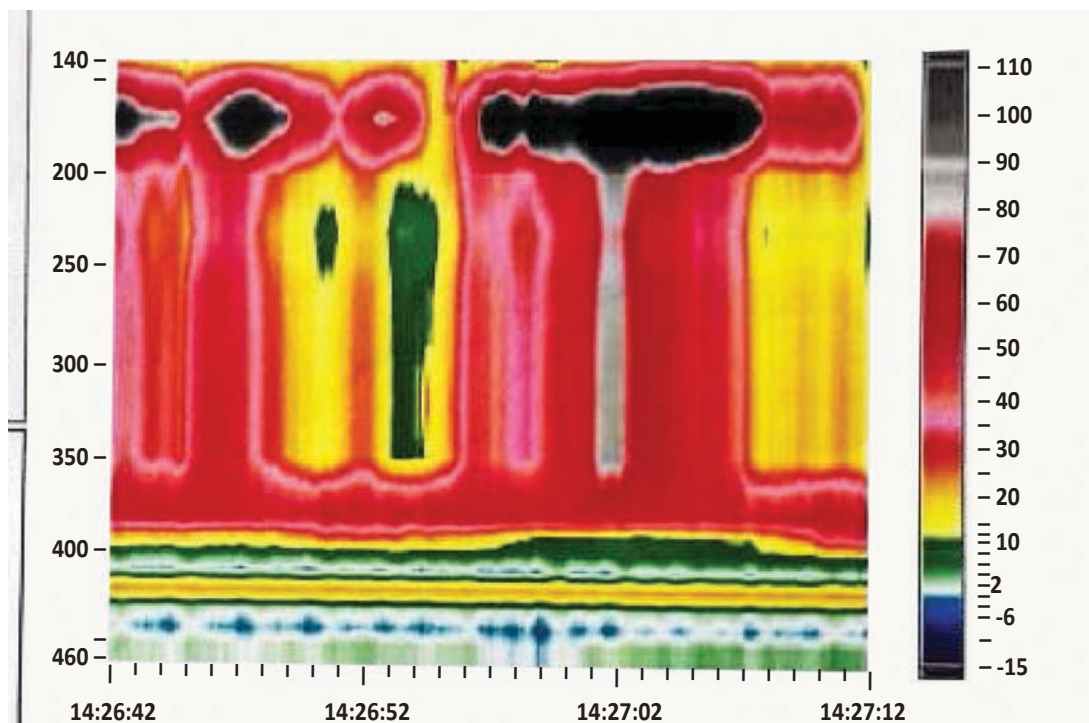


FIGURE 2. Esophageal Manometry This graphic shows pressure as a color gradient (scale on the right), with time on the x-axis and esophageal length on the y-axis (as seen on the left). *High-resolution manometry, which uses more closely spaced sensors than standard manometry, creates this continuous topographic pressure display*

would produce (from top left to bottom right when muscles contract in order), this patient's middle and lower esophageal muscles contract at the same time. There is also no relaxation in the LES.

These results are consistent with Achalasia.

The patient underwent laparoscopic Heller's myotomy and partial fundoplication, along with management for hyperthyroidism. She was started on Tab Carbimazole 10 mg twice daily (for Grave's disease) along with Tab Propranolol 20 mg twice daily (to control her sympathetic symptoms of hyperthyroidism like palpitations), Carbimazole was later tapered to the minimum possible dose to keep her TSH and FT4 within normal limits. The Patient was followed up every 4th week for Thyroid function tests, and to assess her clinical improvement. Following the surgery, she had substantial improvement in her dysphagia, and other symptoms of Hyperthyroidism slowly settled down as her Thyroid functions normalized.

DISCUSSION

Patients presenting with dysphagia are often a challenge when it comes to finding out the cause of the same. Ruling out more common causes like mechanical obstruction, central CNS causes, or local inflammation is a vital part of the evaluation. Neuromuscular disorders causing dysphagia can present with dysphagia to both solids and liquids like in our case. However, an absence of any significant neurological functional deficits and a normal Brain MRI ruled out such a possibility.

Mechanical causes of Dysphagia like esophageal tumours, strictures, webs or rings in the esophagus, and even GERD leading to esophageal strictures can cause dysphagia. Unlike in our case, mechanical causes generally present with dysphagia to solids more than liquids initially and then gradually progress to affect both as the disease progresses. Also, mechanical causes are easily identified by an Upper GI endoscopy, which in our case was not contributory. Extrinsic compression like a mediastinal tumor, lymph nodes, or any mediastinal structure compressing the esophagus can cause dysphagia. These conditions will also have dysphagia to solids first and then to liquids unlike in our case, and may have associated symptoms of the Primary disorder causing compression.

Psychogenic Dysphagia should be considered only after ruling out all other organic causes and hence was not considered in our case. Other systemic disorders like SLE, Rheumatoid arthritis, and Systemic sclerosis can present with similar complaints. Amyloidosis is also an important consideration. The absence of any systemic symptoms for the above conditions and improvement in patient condition following

above mentioned therapies made these differentials an unlikely diagnosis.

Achalasia has been considered a rare disorder, with an annual incidence of roughly 1.6 cases per 100,000 people and a prevalence of 10 cases per 100,000 people [6]. Achalasia is thought to be caused by inflammation and neurodegeneration in the esophageal wall [7]. The findings that achalasia is linked to HLA-DQ region variations and that affected patients frequently have circulating enteric neuron antibodies suggest that achalasia is an autoimmune illness [8]. The degree and location of ganglion cell loss determine the symptoms of achalasia [9]. The most common symptoms in achalasia patients are dysphagia for solids and liquids, as well as regurgitation of bland, partially digested food or saliva [10]. In the following patients, achalasia should be suspected: Dysphagia with regard to solids and liquids, Heartburn that is insensitive to proton pump inhibitor therapy, Upper endoscopy revealed esophageal food retention, Unusual resistance to endoscope passage across the esophagogastric junction. Esophageal manometry is needed to make the diagnosis. Incomplete relaxation of the lower esophageal sphincter (LES; shown as integrated relaxation pressure [IRP] over the upper limit of normal) and a peristalsis in the distal two-thirds of the esophagus are diagnostic manometric findings of achalasia [11]. Achalasia is classified into the following subtypes by the Chicago Classification of Patterns of Esophageal Pressurization on High-resolution manometry; Type I (classic achalasia), Type II (Swallowing causes esophageal pressurization that extends the full length of the esophagus), Type III (spastic achalasia) [12].

Few cross-sectional studies have been done to evaluate the association between thyroid disorders and Achalasia Cardia. One study reported that an association was found between thyroid problems and achalasia and the prevalence of thyroid disease was higher in patients with achalasia than in the general population [4]. The current case describes a patient who arrived with symptoms of achalasia cardia and was also diagnosed with hyperthyroidism. Her symptoms and quality of life improved after laparoscopic Heller's myotomy and partial fundoplication alongside medical management of her hyperthyroidism.

CONCLUSION

Dysphagia for solids and liquids equally along with regurgitation of bland undigested or partially digested food should raise the suspicion of Achalasia Cardia. Having enough evidence for an autoimmune etiology, screening for other autoimmune conditions, which may be coexisting or contributing to achalasia, needs to be done. Although not very common, thyroid disorders can sometimes aggravate the

symptoms of achalasia cardia. Prompt surgical management and treatment of contributing medical conditions can have a substantial impact on the betterment of the quality of life for the patient.

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